

Opis choroby *

Definicja

A rare genetic disorder of lipid metabolism characterized by neonatal to childhood onset of impaired absorption of dietary fat with greasy/oily and voluminous stools, but normal growth and development. Decreased levels of fecal elastase, as well as low serum levels of the fat-soluble vitamins A, D, and E, have been reported.

Dane

Klasyfikacja

Choroba

Synonimy

Pancreatic triglyceride lipase deficiency
Niedobór trzustkowej lipazy trójglicerydów

Kod ORPHA

309031

Kod OMIM

614338

Kod ICD10

K90.3

Kod ICD11

5C62

*Źródło

orphanet